



# GTR: Genetic Testing Registry

Central repository of genetic tests voluntarily supplied by test providers

<https://www.ncbi.nlm.nih.gov/gtr>

National Center for Biotechnology Information • National Library of Medicine • National Institutes of Health • Department of Health and Human Services

## Scope

The Genetic Testing Registry (GTR) is a free online resource that provides centralized access to comprehensive genetic test information voluntarily submitted by test providers. GTR covers clinical and research tests for heritable and somatic mutations, including pharmacogenetic tests as well as tests using complex arrays and multiplex panels. GTR provides a wide range of information, such as, ordering information, purpose of the test and its limitations, testing method(s), and what the test measures. Clinical tests have information on analytical validity, as well as evidence of clinical validity and clinical utility. Research tests have information on the study and participation requirements. The name, location, contact information, and credentials of laboratories are displayed. GTR records provide links to context-specific information about conditions, genes, test standards, practice guidelines, and consumer support sites. The primary audience of GTR is the health care community.



## Data Access

The GTR homepage (right) allows the retrieval of information through several different search strategies using tabs above the search box (A). You can search by the test name, disease, drug response, gene symbol, laboratory name, director and staff names, and laboratory location. A tab for *GeneReviews*, containing reviews on over 700 conditions, enables searching of

this key resource directly from the GTR homepage. A set of YouTube video tutorials (B) provides a quick guide on how to search for and submit information to GTR. Links to GTR documents are in the "About GTR" panel (C). The homepage also provides a summary of participation by laboratories and data growth trends (D). Links to locate a genetics professional, as well as genetics resources incorporated in GTR (E) are provided. The submission section (F) provides links to the submission interface along with help documentation.

<https://www.ncbi.nlm.nih.gov/gtr>

**GTR: GENETIC TESTING REGISTRY**

All GTR Tests Conditions/Phenotypes Genes Labs GeneReviews **A** Advanced search for tests

Search All GTR

Search all 55282 tests, 11276 conditions, 16446 genes, and 507 labs

**IMPORTANT NOTE:** NIH does not independently verify information submitted to the GTR; it relies on submitters to provide information that is accurate and not misleading. NIH makes no endorsements of tests or laboratories listed in the GTR. GTR is not a substitute for medical advice. **Patients and consumers** with specific questions about a genetic test should contact a health care provider or a genetics professional.

**B** YouTube GTR Tutorials

**Medical Genetics Summaries**

Concise reviews about genetic variation and drug responses, accessible at the point of care

Peer reviewed  
Updated regularly  
Freely available  
Standardized format

Therapeutic Recommendations excerpted from FDA, CPIC, & other authoritative source

Variant translation – star alleles, HGVS, etc.

**Find GTR Content**

BRCA1/BRCA2 panels Genomic testing labs  
Mitochondrial genome tests Cancer/somatic tests  
Human genome and Whole exome tests CGH tests  
Pharmacogenetics Single gene tests

All GTR data

**GTR Data**

20K 15K 10K 5K 0 2012 2013 2014 2015 2016 2017 2018

60K 45K 30K 15K 0 2012 2013 2014 2015 2016 2017 2018

Tested Conditions Tested Genes Labs Tests

FTP: Download GTR data and documents

**Locate a Genetics Professional**

ABGC Directory ABMGG Directory NSGC Directory  
NCI Cancer Genetics Services Directory  
ACMG Genetics Clinics Database

**Resources Included in GTR**

GTR includes information from resources such as ClinVar and MedGen from within the NIH and many resources from outside the NIH.  
[See a list of all related resources](#)

**Submitting Information to GTR**

[Access the Submission User Interface](#)  
How to submit data Code of Conduct Submission templates

**About GTR®** **C**

The Genetic Testing Registry (GTR®) provides a central location for voluntary submission of genetic test information by providers. The scope includes the test's purpose, methodology, validity, evidence of the test's usefulness, and laboratory contacts and credentials. The overarching goal of the GTR is to advance the public health and research into the genetic basis of health and disease

- How to use GTR
- Frequently asked questions
- GTR News
- GTR Information at NIH Office of the Director
- GTR in the community
- Contact us and provide feedback

**Worldwide Lab Participation in GTR**

GTR labs by country

**E**

**F**

## Using Advanced Search to Find Specific Tests

You can create custom queries to locate tests using the “Advanced search for tests”. The example (A) shows a search for panels of more than 5 genes for “Primary dilated cardiomyopathy” from a laboratory that provides custom prenatal testing, using method category “Sequence analysis of the entire coding region”. In the search results page, use the preset filters (e.g., “Test purpose”; B) to narrow the list of tests to those fitting the selected criteria (C).

**GTR: GENETIC TESTING REGISTRY**

**Advanced search for tests**  
Find tests that meet all criteria specified below. [Help](#)

Condition name: Primary dilated cardiomyopathy

Number of genes: Greater than 5 less than unlimited

Services: Custom Prenatal Testing

Method category: Enzyme assay

**The search you built**

"Primary dilated cardiomyopathy" AND ("custom prenatal testing") AND Enzyme assay

[Edit](#) [Search](#)

<https://go.usa.gov/xPYEQ>

**Results: 1 to 10 of 10**

Tests names and labs	Conditions	Genes and analytes	Methods
<a href="#">Comprehensive Cardiac Arrhythmia Sequencing Panel with CNV Detection</a> PreventionGenetics United States	68	55	D Deletion/duplication analysis C Sequence analysis of the entire coding region T Targeted variant analysis

**Filters**

**Test type**

☐ Clinical (10)

**Test purpose**

☐ Diagnosis (10)  
☐ Mutation Confirmation (5)  
☐ Pre-symptomatic (2)  
☐ Predictive (1)

**Test method**

**Molecular Genetics**

☐ Deletion/duplication analysis (5)  
☐ Sequence analysis of select exons  
☐ Sequence analysis of the entire coding region (10)  
☐ Targeted variant analysis (1)

**Lab certification**

☐ CLIA Certified (8)  
☐ State Licensed (6)

**Specimen type**

☐ Amniocytes (1)  
☐ Amniotic fluid (1)  
☐ Cell culture (2)  
☐ Chorionic villi (1)  
☐ Fetal blood (1)

**Results: 1 to 5 of 5**

Tests names and labs	Conditions	Genes and analytes	Methods
<a href="#">Comprehensive Cardiac Arrhythmia Sequencing Panel with CNV Detection</a> PreventionGenetics United States	68	55	D Deletion/duplication analysis C Sequence analysis of the entire coding region T Targeted variant analysis
<a href="#">Congenital Myopathy and Distal Myopathy NGS panel</a> Asper Biogene Asper Biogene LLC Estonia	83	41	D Deletion/duplication analysis C Sequence analysis of the entire coding region
<a href="#">Comprehensive Cardiovascular Sequencing Panel</a> EGL Genetic Diagnostics Eurofins Clinical Diagnostics United States	53	117	C Sequence analysis of the entire coding region

## Using the ‘All GTR’ Tab

On the GTR home page, the main search box is preset to the “All GTR” tab. Enter a term (e.g., “Ehlers”) and select from the suggested dropdown list to go to the specific page or click the “Search All GTR” button (D) to get a list of results where you can click the

corresponding buttons to access Tests, Conditions, Genes, and Laboratories (EB). On the Conditions results page, the name of each condition (F) links to the page with condition-specific information (e.g., clinical features, practice guidelines) with links below to available tests, associated genes, and article(s) from *GeneReviews* (GD). Checking the boxes for any conditions activates a link (H) at the top, which can be used to retrieve a subset of tests for any of the selected conditions.

**Ehlers** [Advanced search for tests](#) <https://www.ncbi.nlm.nih.gov/gtr/all/?term=Ehlers>

**Tests (493)** **Conditions (32)** **Genes (22)** **Laboratories (67)**

**Results: 1 to 20 of 32**

2 selected conditions. [Show tests that evaluate any of the checked conditions.](#)

Conditions	Synonyms
<input checked="" type="checkbox"/> <a href="#">Ehlers-Danlos syndrome, type 4</a>	Ehlers Danlos syndrome, Sack-Barabas type Ehlers Danlos syndrome, arterial type Ehlers Danlos syndrome, ecchymotic type Ehlers-Danlos Syndrome Type IV Ehlers-Danlos syndrome vascular type
<input checked="" type="checkbox"/> <a href="#">Ehlers-Danlos syndrome, hydroxylysine-deficient</a>	Cerebral gigantism nevo type EDS VI EHLERS-DANLOS SYNDROME, KYPHOSCOLIOTIC TYPE, 1 EHLERS-DANLOS SYNDROME, OCULAR-SCOLIOTIC TYPE

## Information in a Genetic Test

The example shows a GTR-registered test for warfarin response (<https://www.ncbi.nlm.nih.gov/gtr/tests/568353>) identified by a stable accession and version (A). The test information page opens to show an overview of the test, including the **Purpose of the test**, a **Summary of what is tested**, the **Condition**, and **Genes** involved (B), as well as reported **Clinical Validity** and **Clinical Utility** if relevant and provided (C).

GTR Home > Tests > Warfarin Sensitivity (CYP2C8, CYP2C9, CYP4F2, VKORC1) Genotyping

### Warfarin Sensitivity (CYP2C8, CYP2C9, CYP4F2, VKORC1) Genotyping

Clinical test for Warfarin response  
Offered by ARUP Laboratories, Molecular Genetics and Genomics

GTR Test ID : GTR000568353.1  
Last updated: 2019-07-26  
[Test version history](#)

Test order code : 3001541

**Overview** | How To Order | Indication | Methodology | Performance Characteristics | Interpretation | Laboratory Contact

**Test name**  
Warfarin Sensitivity (CYP2C8, CYP2C9, CYP4F2, VKORC1) Genotyping

**Purpose of the test**  
This is a clinical test intended for : Drug Response, Monitoring, Risk Assessment, Screening, Therapeutic management

**Condition**  
1 condition tested. Click [Indication tab](#) for more information.  
[Warfarin response](#)

**Methodology**  
Molecular Genetics \*  
Sequence analysis of select exons PCR

**Clinical validity**  
Not provided

**Clinical utility**  
Not provided

**How to order**  
Order URL : <https://www.aruplab.com/genetics/ordering>

**Summary of what is tested**  
4 genes and variants. Click [Methodology tab](#) for more information.

**Genes**  
Gene: [CYP2C8](#) (10q23.33)  
Gene: [CYP2C9](#) (10q23.33)  
Gene: [CYP4F2](#) (19p13.12)  
Gene: [VKORC1](#) (16p11.2)

**Reviews**  
Suggested reading  
Furie, 2013  
[WarfarinDosing.org](#)

**Clinical resources**  
MedGen  
OMIM  
PharmGKB  
[Clinicaltrials.gov](#)

**Practice guidelines**  
CPNDS, 2015  
CPIC, 2011  
NACB, 2010  
DailyMed Drug Label, 2010  
ACMG, 2008

**Molecular resources**  
OMIM  
[View CYP2A6 variations in ClinVar](#)  
[View CYP2C9 variations in ClinVar](#)  
[View F9 variations in ClinVar](#)  
[View VKORC1 variations in ClinVar](#)  
RefSeqGene  
Coriell Institute for Medical Research

**Consumer resources**  
Genetic Alliance  
MalaCards  
MedlinePlus

Other information is provided in tabs (D) such as **How to Order** and **Methodology**, which includes the methods and targets that the test interrogates.

Links to important resources (E) are shown on the right. They include **Clinical resources** from NCBI (MedGen and Clinicaltrials.gov) and elsewhere, **Practice guidelines** from professional organizations, as well as **Molecular resources** from NCBI and some outside organizations.

## Information on Testing Laboratories

[GTR Home](#) > [Laboratories](#) > [ARUP Laboratories, Molecular Genetics and Genomics](#)

### ARUP Laboratories, Molecular Genetics and Genomics

ARUP Laboratories, Molecular Genetics and Genomics  
ARUP Laboratories

500 Chipeta Way  
Salt Lake City, Utah, United States 84108  
Phone: 800-242-2787  
Fax: 801-584-5052  
Email: [moleculargcs@aruplab.com](mailto:moleculargcs@aruplab.com)  
Online Contact: <http://www.aruplab.com/genetics/information/contact>  
Website: <http://www.aruplab.com/genetics>

[Submissions in ClinVar](#)  
[Add to preferred labs](#)

<https://www.ncbi.nlm.nih.gov/gtr/>

#### Personnel

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Patti Krautscheid, MS, CGC, Genetic Counselor

#### Conditions and tests 1 tests

[725 conditions/phenotypes with 281 tests](#)

Enter text to narrow down the list

3-Methylglutaconic aciduria type 2

Abacavir hypersensitivity

Achondrogenesis, type IA

Achondrogenesis, type IB

[2 tests](#)

Achondrogenesis, type II

[2 tests](#)

#### List of services

- Custom Deletion/Duplication Testing, [comments](#)
- Identity Testing: Order Code: 0050547
- Mutation Confirmation: Order Code: 2001961, [comments](#)
- Clinical Testing/Confirmation of Mutations Identified Previously: Order Code: 2001961, 2001980, [comments](#)
- Specimen Source Identification: Order Code: 2002067, 2002066, 2002064, 2002065, [comments](#)
- Whole Exome Sequencing: Order Code: 2006332, 2006336
- X-Chromosome Inactivation Studies: Order Code: 2006352

#### List of certifications/licenses

##### Certifications

CLIA, Number: 46D0523979, Expiration date: 2021-02-08

AABB, Number: NA, Expiration date: 2019-12-31

CAP, Number: 4096301, Expiration date: 2019-11-20

ISO15189, Number: NA, Expiration date: 2019-08-19

##### Licenses

CA - California Department of Public Health CDPH, Number: CDS00800007, Expiration date: 2019-12-31

FL - Florida Agency for Health Care Administration AHCA, Number: 800001845, Expiration date: 2019-09-30

MD - Maryland Department of Health and Mental Hygiene DHMH, Number: 516, Effective date: 2018-07-01 Non-expiring

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OR

##### Sign in directly to NCBI

NCBI Username

Password

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Laboratory-specific pages in GTR display the contact information for the lab (A) and personnel (B), tests (C), and services (D) offered by the laboratory as well as CLIA certification and other licenses (E).

A link to ClinVar (F) is provided for participating labs. A link to MyNCBI (G) enables saving of preferred labs and a customized view of their tests.

## Submit Test information to GTR

GTR accepts submissions for laboratory data and clinical and research tests. You will need a MyNCBI account. Detailed instructions are available at [www.ncbi.nlm.nih.gov/gtr/docs/submit/](https://www.ncbi.nlm.nih.gov/gtr/docs/submit/). Submitters may choose to provide their data via:

- Online forms
- Spreadsheets for bulk submissions of clinical tests, either fully automated or semiautomated
- Fully automatic XML submission

GTR is designed to minimize burden on submitters, with features such as menus, "type ahead" functionality, and text fields to allow cut-and-paste of information. Where possible, fields are automatically populated for the submitter. In addition, test data identical for all tests in the lab's menu can be entered in the "Default parameters" section of the lab record to pre-populate fields for new tests being registered.

GTR provides information about the test provider as well as the availability, accuracy, validity and usefulness of each test; therefore, a minimal set of fields are required for all submissions. For the complete list of required and optional fields, see [www.ncbi.nlm.nih.gov/gtr/docs/fieldrequirements/](https://www.ncbi.nlm.nih.gov/gtr/docs/fieldrequirements/). Research tests additionally require information about the study and researchers.

## Contact

GTR welcomes your feedback. Please send questions or comments to: [gtr@ncbi.nlm.nih.gov](mailto:gtr@ncbi.nlm.nih.gov)